



NEB gene

nebulin

Normal Function

The *NEB* gene provides instructions for making a protein called nebulin. This protein plays an important role in skeletal muscles, which are muscles used for movement. Within skeletal muscle cells, nebulin is found in structures called sarcomeres. Sarcomeres are necessary for muscles to tense (contract). Nebulin is one of several proteins that interact to generate the mechanical force needed for muscle contraction.

Health Conditions Related to Genetic Changes

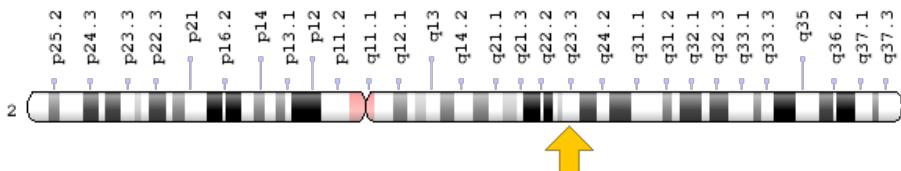
nemaline myopathy

More than 60 mutations in the *NEB* gene have been found to cause nemaline myopathy. Some of these mutations change single protein building blocks (amino acids) in the nebulin protein or result in the production of an abnormally short protein. Other mutations insert or delete genetic material in the *NEB* gene. Most *NEB* gene mutations alter the structure or reduce the production of nebulin. A lack of functional nebulin impairs the muscle cells' ability to contract. Inefficient muscle contraction leads to muscle weakness and the other features of nemaline myopathy.

Chromosomal Location

Cytogenetic Location: 2q23.3, which is the long (q) arm of chromosome 2 at position 23.3

Molecular Location: base pairs 151,485,334 to 151,734,487 on chromosome 2 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- NEBU_HUMAN
- NEM2

Additional Information & Resources

Educational Resources

- Molecular Cell Biology (fourth edition, 2000): Muscle Contraction
<https://www.ncbi.nlm.nih.gov/books/NBK9961/#A1791>
- Molecular Cell Biology (fourth edition, 2000): Titin and Nebulin Filaments Organize the Sarcomere
<https://www.ncbi.nlm.nih.gov/books/NBK21670/#A5209>

GeneReviews

- Nemaline Myopathy
<https://www.ncbi.nlm.nih.gov/books/NBK1288>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28NEB%5BTIAB%5D%29+OR+%28nebulin%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- NEBULIN
<http://omim.org/entry/161650>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_NEB.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=NEB%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=7720
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/4703>
- UniProt
<http://www.uniprot.org/uniprot/P20929>

Sources for This Summary

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- Ottenheijm CA, Hooijman P, DeChene ET, Stienen GJ, Beggs AH, Granzier H. Altered myofilament function depresses force generation in patients with nebulin-based nemaline myopathy (NEM2). *J Struct Biol.* 2010 May;170(2):334-43. doi: 10.1016/j.jsb.2009.11.013. Epub 2009 Nov 26.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/19944167>
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- Ryan MM, Schnell C, Strickland CD, Shield LK, Morgan G, Iannaccone ST, Laing NG, Beggs AH, North KN. Nemaline myopathy: a clinical study of 143 cases. *Ann Neurol.* 2001 Sep;50(3):312-20. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/11558787>

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